(FILE 'HOME' ENTERED AT 13:20:48 ON 21 MAR 2001)

FILE 'CAPLUS, USPATFULL, WPIDS, DGENE, EUROPATFULL, JAPIO, MEDLINE'
ENTERED AT 13:21:31 ON 21 MAR 2001

| | | | |
|---|---|---|---|
| L1 | 45 | S | MAE (W) II |
| L2 | 1 | S | L1 AND DPD |
| L3 | 2350 | S | DPD |
| L4 | 104 | S | L3 AND (MUTATION OR POLYMORPHISM) |
| L5 | 44 | S | L4 NOT PY>1996 |
| L6 | 0 | S | 1-10 IBIB ABS |

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=> d his

     (FILE 'HOME' ENTERED AT 14:07:01 ON 16 MAR 2001)

     FILE 'CAPLUS, USPATFULL, WPIDS, DGENE, EUROPATFULL, JAPIO' ENTERED AT
     14:07:34 ON 16 MAR 2001
L1           353 S DIHYDROPYRIMIDINE (W) DEHYDROGENASE
L2        288659 S MUTATION OR POLYMORPHISM
L3            71 S L2 AND L1
L4             1 S L3 AND MAEIII
L5             0 S L1 AND ALTERNATE (W) SPLIC?
L6            41 S L1 AND SCREEN?

=> s l6 not py>1996

   3 FILES SEARCHED...
L7             9 L6 NOT PY>1996

=> d ibib abs 1-

YOU HAVE REQUESTED DATA FROM 9 ANSWERS - CONTINUE? Y/(N):y


L7   ANSWER 1 OF 9  CAPLUS  COPYRIGHT 2001 ACS
ACCESSION NUMBER:         1996:690425  CAPLUS
DOCUMENT NUMBER:          125:325384
TITLE:                    A point mutation in an invariant splice donor site
                          leads to exon skipping in two unrelated Dutch patients
                          with dihydropyrimidine dehydrogenase
                          deficiency
AUTHOR(S):                Vreken, P.; Van Kuilenburg, B. P.; Meinsma, R.; Smit,
                          G. P. A.; Bakker, H. D.; De Abreu, R. A.; van Gennip,
                          A. H.
CORPORATE SOURCE:         Acad. Med. Cent., Univ. Amsterdam, Amsterdam, 1100 DE,
                          Neth.
SOURCE:                   J. Inherited Metab. Dis. (1996), 19(5), 645-654
                          CODEN: JIMDDP; ISSN: 0141-8955
DOCUMENT TYPE:            Journal
LANGUAGE:                 English
AB   Dihydropyrimidine dehydrogenase (DPD) deficiency is an
     autosomal recessive disease characterized by thymine-uraciluria and
     assocd. with a variable clin. phenotype.  To identify the mol. defect
     underlying complete DPD deficiency in a Dutch patient previously shown to
     have a 165 base pair deletion in the mature DPD mRNA, the authors cloned
     the genomic region encompassing the skipped exon and its flanking intron
     sequences.  Sequence anal. revealed that the patient was homozygous for a
     single G .fwdarw. A point mutation in the invariant GT dinucleotide splice
     donor site downstream of the skipped exon.  The same mutation was
     identified in another, unrelated, Dutch patient.  Because this mutation
     destroys a unique MaeII restriction site, rapid screening using
     restriction enzyme cleavage of the amplified genomic region encompassing
     this mutation is possible.  Anal. of 50 controls revealed no individuals
     heterozygous for this mutation.

L7   ANSWER 2 OF 9  CAPLUS  COPYRIGHT 2001 ACS
ACCESSION NUMBER:         1996:483095  CAPLUS
DOCUMENT NUMBER:          125:139772
TITLE:                    Molecular basis of the human dihydropyrimidine
                          dehydrogenase deficiency and 5-fluorouracil
                          toxicity
AUTHOR(S):                Wei, Xiaoxiong; McLeod, Howard L.; McMurrough,
                          Julieann; Gonzalez, Frank J.; Fernandez-Salguero,
                          Pedro
CORPORATE SOURCE:         Laboratory of Molecular Carcinogenesis, National
                          Institutes of Health, Bethesda, MD, 20892, USA
SOURCE:                   J. Clin. Invest. (1996), 98(3), 610-615
                          CODEN: JCINAO; ISSN: 0021-9738
DOCUMENT TYPE:            Journal
LANGUAGE:                 English
AB   Dihydropyrimidine dehydrogenase (DPD) deficiency
     constitutes an inborn error in pyrimidine metab. assocd. with
     thymine-uraciluria in pediatric patients and an increased risk of toxicity
     in cancer patients receiving 5-fluorouracil (5-FU) treatment.  The mol.
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